Your Guide to Understanding Genetic Conditions

OSMR gene

oncostatin M receptor

Normal Function

The OSMR gene provides instructions for making a protein called oncostatin M receptor beta subunit ($OSMR\beta$). This protein is one piece (subunit) of both the oncostatin M (OSM) receptor type II and the interleukin-31 (IL-31) receptor. These receptors are embedded in the cell membrane of many types of cells throughout the body. Each attaches to a particular protein, fitting together like a lock and its key. This attachment triggers a series of chemical signals inside the cell that directs certain cell functions.

OSM receptor type II interacts with a protein called oncostatin M (OSM). Signaling triggered by OSM was first recognized to block the growth of cancerous cells and appears to play a role in many other body processes, including the development of blood cells, the maturation of cells to become certain cell types, and an immune system response called inflammation. The signaling may also block the self-destruction (apoptosis) of cells.

The IL-31 receptor interacts with a protein called IL-31. Signaling triggered by IL-31 is involved in inflammation and stimulating itching (pruritus), although its role is not completely understood.

Health Conditions Related to Genetic Changes

primary localized cutaneous amyloidosis

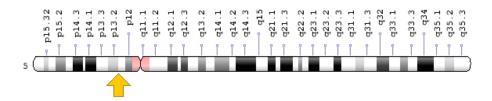
At least 13 mutations in the *OSMR* gene have been found to cause primary localized cutaneous amyloidosis (PLCA) type 1, an itchy skin condition in which clumps of abnormal proteins called amyloids build up in the skin. These mutations change single protein building blocks (amino acids) in OSMRβ. Most alter a region of the protein thought to interact with the other subunit of the OSM receptor type II or the IL-31 receptor and may impair formation of these receptors.

OSMR gene mutations reduce the chemical signals triggered by OSM and IL-31. Researchers speculate that this reduced signaling may make cells more likely to undergo apoptosis. Some studies suggest that apoptosis of skin cells releases abnormal proteins that form amyloids. It has been suggested that apoptosis is triggered by scratching the itchy skin, but the role of OSMR gene changes in skin itching is not clear.

Chromosomal Location

Cytogenetic Location: 5p13.1, which is the short (p) arm of chromosome 5 at position 13.1

Molecular Location: base pairs 38,845,858 to 38,945,579 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- IL-31 receptor subunit beta
- IL-31R-beta
- IL-31R subunit beta
- IL-31RB
- interleukin-31 receptor subunit beta
- oncostatin-M specific receptor beta subunit
- OSMRB
- PLCA1

Additional Information & Resources

Educational Resources

- Itch: Mechanisms and Treatment (2014): Role of Interleukin-31 and Oncostatin M in Itch and Neuroimmune Communication
 https://www.ncbi.nlm.nih.gov/books/NBK200913/
- Molecular Biology of the Cell (fourth edition, 2002): General Principles of Cell Communication https://www.ncbi.nlm.nih.gov/books/NBK26813/

Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28OSMR%5BTIAB %5D%29+OR+%28oncostatin+M+receptor%5BTIAB%5D%29%29+AND+ %28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D %29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last +1800+days%22%5Bdp%5D

OMIM

 ONCOSTATIN M RECEPTOR http://omim.org/entry/601743

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology http://atlasgeneticsoncology.org/Genes/GC_OSMR.html
- ClinVar https://www.ncbi.nlm.nih.gov/clinvar?term=OSMR%5Bgene%5D
- HGNC Gene Family: Fibronectin type III domain containing http://www.genenames.org/cgi-bin/genefamilies/set/555
- HGNC Gene Symbol Report http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/ hgnc_data.php&hgnc_id=8507
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/9180
- UniProt http://www.uniprot.org/uniprot/Q99650

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